



We Claim:

1. An isolated nucleic acid comprising any one of SEQ ID NOS:1-30, or a sequence complementary to any one of SEQ ID NOS:1-30.
2. An isolated nucleic acid comprising at least eight consecutive nucleotides of a nucleotide sequence of any one of SEQ ID NOS:1-30, or at least eight consecutive nucleotides of a nucleotide sequence complementary to any one of SEQ ID NOS:1-30.
3. An isolated nucleic acid comprising at least 80% nucleotide identity with a nucleic acid comprising any one of SEQ ID NOS:1-30, or at least 80% nucleotide identity with a sequence complementary to any one of SEQ ID NOS:1-30.
4. The isolated nucleic acid according to claim 3, wherein the nucleic acid comprises at least an 85%, 90%, 95%, or 98% nucleotide identity with a nucleic acid comprising any one of SEQ ID NOS:1-30, or comprises at least an 85%, 90%, 95%, or 98% nucleotide identity with a sequence complementary to any one of SEQ ID NOS:1-30.
5. An isolated nucleic acid that hybridizes under high stringency conditions with a nucleic acid comprising any one of SEQ ID NOS:1-30, or with a nucleic acid comprising a nucleotide sequence complementary to any one of SEQ ID NOS:1-30..
6. A nucleotide probe or primer specific for an ABCC11 gene, wherein the nucleotide probe or primer comprises at least 15 consecutive nucleotides of a nucleotide



sequence of any one of SEQ ID NOS:1-30, or at least 15 consecutive nucleotides of a sequence complementary to any one of SEQ ID NOS:1-30.

7. A nucleotide probe or primer specific for an ABCC11 gene, wherein the nucleotide probe or primer comprises a nucleotide sequence of any one of SEQ ID NOS:1-30, or a nucleotide sequence complementary to any one of SEQ ID NOS:1-30.
8. A method of amplifying a region of the nucleic acid according to claim 1, comprising:
  - a) contacting the nucleic acid with two nucleotide primers, wherein the first nucleotide primer hybridizes at a position 5' of the region of the nucleic acid to be amplified, and the second nucleotide primer hybridizes at a position 3' of the region of the nucleic acid to be amplified, in the presence of reagents necessary for an amplification reaction; and
  - b) amplifying the nucleic acid region; and
  - c) detecting the amplified nucleic acid region.
9. The method according to claim 8, wherein each nucleic acid primer is independently selected from the group consisting of
  - a) a nucleotide primer comprising at least 15 consecutive nucleotides of a nucleotide sequence of any one of SEQ ID NOS:1-30,
  - b) a nucleotide primer comprising at least 15 consecutive nucleotides of a nucleotide sequence complementary to any one of SEQ ID NOS:1-30,
  - c) a nucleotide primer as in any one of claims 6-8,

d) a nucleotide primer comprising a nucleotide sequence of any one of SEQ ID NOS:1-30, and

e) a nucleotide primer comprising a nucleotide sequence complementary to any one of SEQ ID NOS:1-30.

10. A kit for amplifying the nucleic acid according to claim 1, comprising:

a) two nucleotide primers whose hybridization position is located respectively 5' and 3' of the region of the nucleic acid to be amplified; and optionally,

b) reagents necessary for an amplification reaction.

11. The kit according to claim 10, wherein each nucleic acid primer is independently selected from the group consisting of

a) a nucleotide primer comprising at least 15 consecutive nucleotides of a nucleotide sequence of any one of SEQ ID NOS:1-30,

b) a nucleotide primer comprising at least 15 consecutive nucleotides of a nucleotide sequence complementary to any one of SEQ ID NOS:1-30,

c) a nucleotide primer as in any one of claims 6-8,

d) a nucleotide primer comprising a nucleotide sequence of any one of SEQ ID NOS:1-30, and

e) a nucleotide primer comprising a nucleotide sequence complementary to any one of SEQ ID NOS:1-30.

12. The nucleotide probe or primer according to any one of claims 6-8, wherein the nucleotide probe or primer comprises a marker compound.



13. A method of detecting a nucleic acid according to claim 1, comprising:
- a) contacting the nucleic acid to be detected with a nucleotide probe selected from the group consisting of
    - i) a nucleotide primer comprising at least 15 consecutive nucleotides of a nucleotide sequence of any one of SEQ ID NOS:1-30,
    - ii) a nucleotide primer comprising at least 15 consecutive nucleotides of a nucleotide sequence complementary to any one of SEQ ID NOS:1-30,
    - iii) a nucleotide primer as in any one of claims 6-8,
    - iv) a nucleotide primer comprising a nucleotide sequence of any one of SEQ ID NOS:1-30, and
    - v) a nucleotide primer comprising a nucleotide sequence complementary to any one of SEQ ID NOS:1-30; and
  - b) detecting a complex formed between the nucleic acid and the probe.
14. The method of claim 13, wherein the probe is immobilized on a support.
15. A kit for detecting the nucleic acid according to claim 1, wherein the kit comprises
- a) a nucleotide probe selected from the group consisting of
    - i) a nucleotide primer comprising at least 15 consecutive nucleotides of a nucleotide sequence of any one of SEQ ID NOS:1-30,

ii) a nucleotide primer comprising at least 15 consecutive nucleotides of a nucleotide sequence complementary to any one of SEQ ID NOS:1-30,

iii) a nucleotide primer as in any one of claims 6-8,

iv) a nucleotide primer comprising a nucleotide sequence of any one of SEQ ID NOS:1-30, and

v) a nucleotide primer comprising a nucleotide sequence complementary to any one of SEQ ID NOS:1-30; and optionally,

b) reagents necessary for a hybridization reaction.

16. The kit according to claim 15, wherein the probe is immobilized on a support.

17. A recombinant vector comprising the nucleic acid according claim 1.

18. The vector according to claim 17, wherein the vector is an adenovirus.

19. A recombinant host cell comprising the recombinant vector according to claim 17.

20. A recombinant host cell comprising the nucleic acid according claim 1.

21. An isolated nucleic acid encoding a polypeptide comprising an amino acid sequence of SEQ ID NO:31.

22. A recombinant vector comprising the nucleic acid according to claim 21.



23. A recombinant host cell comprising the nucleic acid according to claim 21.
24. A recombinant host cell comprising the recombinant vector according to claim 22.
25. An isolated polypeptide selected from the group consisting of
- a) a polypeptide comprising an amino acid sequence of SEQ ID NO:31,
  - b) a polypeptide fragment or variant of a polypeptide comprising an amino acid sequence of SEQ ID NO:31, and
  - c) a polypeptide homologous to a polypeptide comprising an amino acid sequence of SEQ ID NO:31.
26. An antibody directed against the isolated polypeptide according to claim 25.
27. The antibody according to claim 26, wherein the antibody comprises a detectable compound.
28. A method of detecting a polypeptide, comprising:
- a) contacting the polypeptide with an antibody according to claim 26; and
  - b) detecting an antigen/antibody complex formed between the polypeptide and the antibody.
29. A diagnostic kit for detecting a polypeptide, comprising:
- a) the antibody according to claim 26; and



b) a reagent allowing detection of an antigen/antibody complex formed between the polypeptide and the antibody.

30. A pharmaceutical composition comprising the nucleic acid according to claim 1 and a physiologically compatible excipient.

31. A pharmaceutical composition comprising the recombinant vector according to claim 17 and a physiologically compatible excipient.

32. A method of treating and/or preventing paroxysmal kinesigenic choreoathetosis in a subject in need thereof by administering the nucleic acid according to claim 1.

33. A method of treating and/or preventing paroxysmal kinesigenic choreoathetosis in a subject in need thereof by administering the recombinant vector according to claim 20.

34. A method of treating and/or preventing paroxysmal kinesigenic choreoathetosis in a subject in need thereof by administering an isolated ABCC11 polypeptide comprising the amino acid sequence of SEQ ID NO:31.

35. A pharmaceutical composition comprising a polypeptide comprising an amino acid sequence of the SEQ ID NO:31, and a physiologically compatible excipient.

36. A method of identifying active ingredients for the prevention or treatment of paroxysmal kinesigenic choreoathetosis using an isolated ABCC11 polypeptide comprising an amino acid sequence of SEQ ID NO:31

37. A method of identifying active ingredients for the prevention or treatment of paroxysmal kinesigenic choreoathetosis using a recombinant host cell expressing an ABCC11 polypeptide comprising an amino acid sequence of SEQ ID NO:31.

38. A method of screening an agonist or an antagonist of an ABCC11 polypeptide, comprising:

- a) preparing a membrane vesicle comprising at least one of the ABCC11 polypeptide and a substrate comprising a detectable marker;
- b) incubating the vesicle obtained in step a) with an agonist or antagonist candidate compound;
- c) qualitatively and/or quantitatively measuring a release of the substrate comprising the detectable marker; and
- d) comparing the release of the substrate measured in step b) with a measurement of a release of a labelled substrate by a membrane vesicle that has not been previously incubated with the agonist or antagonist candidate compound.

39. A method of screening an agonist, or an antagonist of an ABCC11 polypeptide, comprising

- a) incubating a cell that expresses the ABCC11 polypeptide with an anion labelled with a detectable marker;



b) washing the cell

of step a) whereby excess labelled anion that has not penetrated into the cell is removed;

c) incubating the cell obtained in step b) with an agonist or antagonist candidate compound for the ABCC11 polypeptide;

d) measuring efflux of the labelled anion from the cell; and

e) comparing the efflux of the labelled anion determined in step d) with efflux of a labelled anion measured with a cell that has not been previously incubated with the agonist or antagonist candidate compound.

40. An implant comprising the recombinant host cell according to claim 24 or 25.